ANK1 gene

ankyrin 1

Normal Function

The *ANK1* gene provides instruction for making a protein called ankyrin-1. This protein is primarily active (expressed) in red blood cells, but it is also found in muscle and brain cells. In red blood cells, ankyrin-1 is located at the cell membrane, where it attaches (binds) to other membrane proteins. The binding of membrane proteins to one another maintains the stability and structure of red blood cells but also allows for their flexibility. The proteins allow the cell to change shape without breaking when passing through narrow blood vessels.

In muscle and brain cells, ankyrin-1 performs similar functions, binding to other membrane proteins to play a role in cell stability, cell movement, and other cell functions.

Health Conditions Related to Genetic Changes

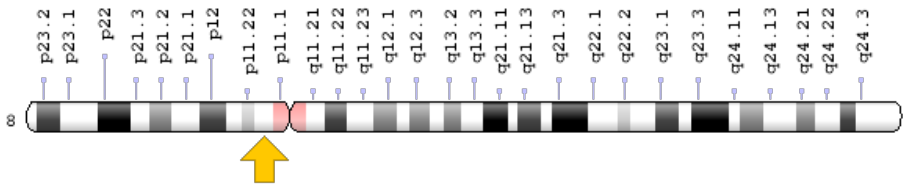
hereditary spherocytosis

At least 55 mutations in the *ANK1* gene have been found to cause hereditary spherocytosis. Some of these mutations delete small pieces of genetic material, and others change single DNA building blocks (nucleotides) in the *ANK1* gene. These mutations lead to the production of an ankyrin-1 protein that does not function normally and does not bind to other proteins within the red blood cell membrane. A lack of normal ankyrin-1 at the cell membrane also leads to a lack of another protein called spectrin because ankyrin-1 is not available to bind to spectrin. The shortage (deficiency) of these two proteins interferes with the structure and flexibility of the red blood cell membrane, causing red blood cells to be misshapen. These misshapen cells, called spherocytes, are removed from circulation and taken to the spleen for destruction. The shortage of red blood cells in circulation and the abundance of cells in the spleen are responsible for the signs and symptoms of hereditary spherocytosis.

Chromosomal Location

Cytogenetic Location: 8p11.21, which is the short (p) arm of chromosome 8 at position 11.21

Molecular Location: base pairs 41,653,225 to 41,896,762 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ANK
- ANK-1
- ANK1_HUMAN
- ankyrin-1
- ankyrin 1, erythrocytic
- ankyrin-R
- erythrocyte ankyrin

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Erythrocyte Cytoskeleton https://www.ncbi.nlm.nih.gov/books/NBK21493/

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ANK1%5BTIAB%5D%29+OR+%28ankyrin-R%29%29+OR+%28ankyrin-1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- ANKYRIN 1
  http://omim.org/entry/612641

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ANK1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ANK1%5Bgene%5D
- HGNC Gene Family: Ankyrin repeat domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/403
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=492
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/286
- UniProt
  http://www.uniprot.org/uniprot/P16157

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